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Spine ultrasound evaluation in occult spinal dysraphism: A 5-year retrospective observation

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Abstract

Aim: Spine ultrasound scans are commonly used in neonates and infants with sacral dimples to rule out associated congenital malformations. In this study, we aimed to evaluate the type and frequency of spinal dysraphisms in patients examined by superficial spine ultrasound, which is used in screening vertebral and spinal anomalies.

Material and Methods: We retrospectively reviewed the presence of suspicious lumbosacral skin manifestations and Spine ultrasound findings in our pediatric clinic over a 4-year period (June 2015-December 2019). The patients' natal histories, comorbidities, sacral physical examination findings, and spine ultrasound scans were evaluated from the medical records.

Results: Among n=1854 spine ultrasounds, n=1708 were included in the study. On physical examination, there were 56.5% sacral dimples, 19.1% bifurcations, 18.7% sacral hair growth, 3.5% hemangiomas or discolorations, 1.5% sacral sinus, 0.5% skin tag, and 0.1% sacral asymmetry. Multiple examination findings were present in 6.3% of the patients. In spine ultrasounds, most reports had normal (90.2%) or variants of normal findings (8.5%). 7.6% (n=130) filar cysts and 0.5%(n=9) terminal ventricles were reported. The number of patients with pathological findings in spinal ultrasound results was n=23 (1.3%). Tethered cord was reported in 0.6% (n=10), syringomyelia in 0.5% (n=8), diastomyelia in 0.1% (n=2), and solid mass in 0.1% (n=2) and sacral agenesis in 1 patient.

Discussion: In healthy infants with sacral dimples, the risk of major spinal dysraphism is exceedingly minimal. Spinal ultrasonographic scanning is a simple and safe tool for patients with suspected spinal dysraphism and sacral skin findings. Clinical suspicion is still of primary importance in recognizing conditions such as tethered cord syndrome, which are rare but can contribute significantly when diagnosed early.

Keywords

Infant, Occult Spinal Dysraphism, Spine Ultrasound

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Introduction

Occult Spinal Dysraphism (OSD) means incomplete closure of midline structures and anomalous improvement of the caudal cell mass at some point of embryogenesis. Its frequency varies depending on ethnicity, geography, and gender [1].

Some skin findings in the lumbosacral region may accompany OSD; these are hemangiomas, increased hair growth (hypertrichosis), skin folds (skin tag), sacral dimple, dermal sinus, subcutaneous mass, curvature in the gluteal cleft, aplasia cutis [2]. Lumbosacral cutaneous manifestations (LsCMs) are seen in 51% to 100% of children with OSDs [3,4]. The relationship between simple sacral dimples and tethered cord syndrome or other spine anomalies is not well understood. One type of OSD, tethered cord syndrome is associated with congenital malformations and some cutaneous stigmata, may cause neurological, urological, and orthopedic sequelae [5]. Examination of the child with further investigation allows to to establish whether the visual abnormalities can be completely distinguished via visual examination [6]. Before developing neurological findings in these patients, early diagnosis of OSD for early surgical intervention recognition is essential [7]. Ultrasound and magnetic resonance imaging are the preferred imaging modalities for spine examinations in newborns up to 3 months of age [8]. Ulrasound is a safe, non-invasive imaging modality that does not require sedation. It does not use radiation, and parents widely accept its results. Consequently, this procedure is well established to investigate the spinal cord and canal in pregnancy and newborns to recognize spinal abnormalities. Ultrasound imaging method is an effective screening tool for the presence of a tethered cord, with a sensitivity of 96% and a specificity of 96% [9,10].

In this study, we discuss the identification of lumbosacral cutaneous manifestations that predict OSD. We retrospectively analyzed 1854 spine ultrasounds performed in our hospital between 01 June 2015 and 30 June 2019 and evaluated 1708 cases with skin findings in terms of OSD and compared them with the literature.

Material and Methods

We reviewed 1854 cases with suspicious skin findings at the lumbar spinal region and spine ultrasounds performed in Zeynep Kamil Maternity and Child Health and Diseases Hospital retrospectively from June 2015 through December 2019. One thousand seven hundred and eight cases were included in the study and evaluated from the electronic data of the hospital. The ultrasound findings of the patients who underwent the first ultrasound within three months after their birth were analyzed. Ultrasound of the lumbar spine is generally not performed at our institutions after three months due to ossification of the posterior elements of the spine by this age.

Data collection and flowchart are presented in Figure 1.

Cases Inclusion criteria:

1. 0-3 months age
2. Suspicious skin findings in terms of OSD
3. A superficial ultrasonographic imaging of the spinal region was performed

Criteria for exclusion of cases from the study:

1. Age older than three months
2. No superficial ultrasonographic imaging of the spinal region
3. Previously diagnosed with other medical conditions such as neurological diseases, congenital abnormalities, syndromes.
4. Grossly observed anal anomalies and multiple deformities.

Gestational age, gender, and skin findings in the spinal region detected on physical examination were recorded.

After superficial ultrasonography for further examination, all patients with abnormal spine ultrasound results were referred to a pediatric neurosurgeon, who determined whether a lumbar MRI was necessary.

This retrospective chart review study involving human participants was conducted in accordance with the ethical standards of the institutional and national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. The local Ethics Committee approved this study (no: 121, date: November 09, 2022).

Statistical analysis

Windows-based SPSS 26.0 analysis program was used for statistical analysis. Normality of the data was examined with the Kolmogorov-Smirnov test. In the evaluation of the data, descriptive statistics were used for demographic data. Mean ± standard deviation and median (min-max values calculated for variables) were determined by measuring within the counted variables (%), and frequency values were calculated. The chi-square test was used to compare the qualitative data. $p < 0.05$ was considered to indicate statistical significance.

Ethical Approval

Ethics Committee approval for the study was obtained.

Results

From June 2015 to December 2019, 1854 cases who applied to our hospital with the complaint of skin findings or who had suspicious skin findings during the examination were retrospectively reviewed. One hundred fifty of 1854 cases older than three months or with incorrect ultrasound requests were not included in the study. The flow diagram for enrollment was drawn in Figure 1. Of the 1708 patients included in the study, 51.1% were boys, and 48.9% were girls. The mean gestational age was 263±16.4 days, and birth weight was 3149.5 ±567.7 gr. The median age at admission was 28 (0-90) days (Table

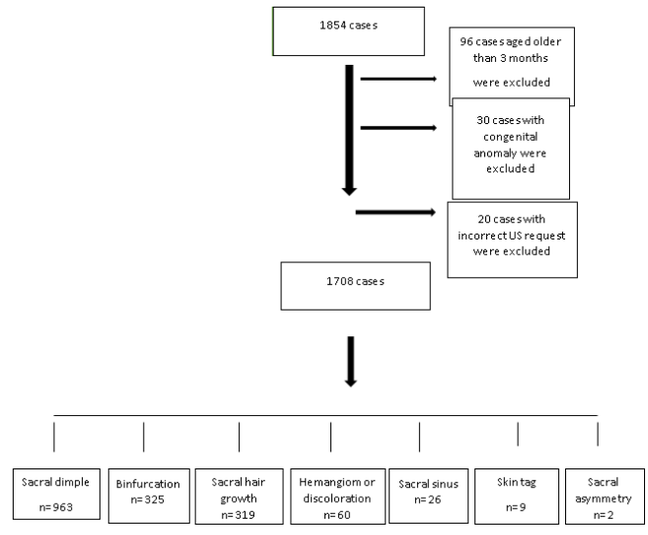

Figure 1. Flow chart of the study.

Table 1. Baseline characteristics of patients

Patient profiles	
Variable	**Value**
Demographic findings	
Sex, male: female	873:835
Age at visit (day)	28 (0–90)*
Delivery history	
Gestational age (day)	263±16.4•
Preterm (<37)	485 (%28.4)
Term (≥37, <42)	1218 (%71.3)
Postterm (≥42)	5 (%0.3)
Birth weight (gr)	3149.5±567.7•
Delivery method, VD: C/S	1006:702 (%58.9: %41.1)

*median value; •mean±sd

1). The main reasons for admission were routine well-child visits (53.5% (n=909)) and hip dislocation screening (29.7% (n=505)). Only 16.7% (n=284) of the patients had active complaints. Of these complaints, 25.3% (n=72) were related to spinal dysraphism. On physical examination, there were 56.5% sacral dimples, 19.1% bifurcations, 18.7% sacral hair growth, 3.5% hemangiomas or discolorations, 1.5% sacral sinus, 0.5% skin tag, and 0.1% sacral asymmetry (Figure 1). Multiple examination findings were present in 6.3% of the patients. In spinal USGs, most reports had normal (90.2%) or a variant of normal (8.5%) findings. 7.6% (n=130) filar cysts and 0.5% (n=9) terminal ventricles were reported. The number of patients with pathological findings in spinal USG results was n=22 (1.3%). Tethered cord was reported in 0.6% (n=10), syringomyelia in 0.5% (n=8), diastomyelia in 0.1% (n=2), and solid mass in 0.1% (n=2) of patients, sacral agenesis in 1 patient. No statistical significance was found between gender (p=0.343), low birth weight (p=0.262), preterm birth (p=0.051), presence of multiple examination findings (p=0.313), and spinal pathologies. All patients (n=22) with tethered cord, syringomyelia, diastomyelia, and suspected mass were referred to neurosurgery. It was seen that 16.9% of the patients with a variant of normal findings were referred to neurosurgery.

Discussion

Although there is no consensus on imaging in the literature, it is important to determine whether even a single patient can be performed surgical correction. Although occult spinal dysraphism's frequency varies in different studies, the fact that patients who cannot be diagnosed early may experience urological, neurological and, orthopedic sequelae and meningitis in the future shows the importance of imaging. In the lumbosacral region of an infant, a wide spectrum of cutaneous findings may be observed, with varying degrees of suspicion for OSD. In the scientific literature, 80% of patients diagnosed with tethered cord syndrome have evidence of midline cutaneous lesions over the lumbosacral spine [4,11].

Shields LB et al. reported in their study that, due to the risk of specific lumbosacral cutaneous manifestations with occult spinal dysraphism, all lumbosacral cutaneous manifestations in patients with a suspected spinal dysraphism should be investigated using a spine ultrasound [12].

Features considered high-risk for OSD include dimpling and mass in the lumbar region, large pedunculated lesions, raised hemangiomas, dermal sinus tract, subcutaneous lipoma, and caudal appendage. Low-risk features include a flat hemangioma, non-midline lesion (such as a forked gluteal cleft), coccygeal pit, or simple sacral dimple [11,13]. In our study, the most common skin finding was sacral dimple (56.5%). Kriss et al. defined 'atypic sacral dimples as more than 5 mm in depth and 2.5 cm in height from the anus, requiring imagining, and found the frequency of sacral dimples to be 74% in their study [14]. We saw that a spinal abnormality was detected in only 14 patients (1,2%) with sacral dimples, and referred to neurosurgery. In the literature review, the general opinion is that further investigation is unnecessary for an isolated sacral dimple. Kucera NJ et al. reported that in asymptomatic infants with an isolated sacral dimple, the risk of spinal malformations is exceedingly low. From their large cohort of asymptomatic children with simple sacral dimples, only 0.13% led to surgery [15]. Albert GW reported that sacral dimples do not indicate a spinal cord malformation and that a spine ultrasound should not be performed on neonates with simple sacral dimples. The review found that the incidence of spinal abnormalities in children with simple sacral dimples (3.4% was very close to that of children without sacral dimples (4.8%) [16]. Hypertrichosis is commonly observed with other cutaneous manifestations of OSD, which indicate an underlying spinal defect. The association of increased hair growth in the sacral region with diastematomyelia is known [17]. McGovern et al. [18] reported a hair growth rate of 4% over 216 cases, and a hair growth rate of 14% in the cases with 144 skin findings was found in a study by Henriques et al. In both studies, diastematomyelia was not accompanied by hair growth [19].

In our study, hair growth was found in 18.7 %, and 3 (0,94 %) patients had spinal abnormalities.

The presence of more than one skin finding is considered more significant in terms of OSD. In the literature, McGovern et al. have shown that OSD detection of the presence of multiple skin findings was six times more significant than a single skin finding [18].

A review of 54 cases of congenital midline lumbosacral cutaneous lesions observed by a pediatric dermatologist, detected OSD in 11 of 18 patients with two or more different skin lesions [20].

The most common complex skin manifestations are associated with sacral dimples and hair growth [2,18]. In our study, the frequency of complex skin findings was 0,31%, and the most common complex finding was sacral dimple and hair growth; none of these cases had OSD.

A spine ultrasound is a simple, inexpensive, and noninvasive screening tool for detecting OSD of the vertebrae in infants less than three months old when the bones have not yet been fully ossified [8]. A patient with an abnormal or equivocal spine, or one that was limited due to ossification of the spine, should undergo magnetic resonance imaging (MRI). The MRI is the most sensitive imaging modality for detecting osteosclerosis in infants. However, its use comes with several drawbacks: it entails the need for general anesthesia, which has its own risks and costs more than other methods; it is more difficult

to obtain; availability may be limited, and infants' heartbeats can generate motion that can complicate image acquisition [21]. Shields LB. et al. [12] recommended if a patient is younger than three months, undergo the US; if the patient is older than six months, MRI without contrast; and if a patient meets the criteria for neurosurgery, they recommend referral to pediatric neurosurgery. In our study, we reviewed the patients' spine ultrasound findings for three months and, showed that all patients with pathological findings were referred to pediatric neurosurgery. According to Irani et al. [22] filar cysts occurred at a frequency of 11.8 %, and the short-term outcomes were not significantly different from those of normal controls. Neonatal filar cysts found isolated on lumbar ultrasound can be considered normal variants, despite the fact that little is known about how and why filar cysts exist. In our study, the incidence of filar cysts was 7.6%. However, interestingly, it was seen that 16.9% of the patients with a variety of normal findings were referred to neurosurgery. A neonatal spine ultrasound can demonstrate several spinal variants. Common spinal cord variants include ventriculus terminalis, filar cyst, thickened filum terminale and a fibrous tract from the coccyx [23]. These should be known to the sonographer and clinician not to cause unnecessary additional imaging or stress for parents or guardians.

Limitation

The limitations of this study were that it was a retrospective, single-institutional study. Secondly, we need to find out the follow-up of children with pathological USG findings. Future prospective studies should be conducted to evaluate long-term clinical outcomes. Third, because of the retrospective nature of this study, medical records were insufficient, and we could not classify the sacral dimples. Because of the retrospective nature of this study, the authors were unable to examine the children's sacral dimples, without determining whether the sacral dimple was simple in nature or complex.

Conclusion

Although OSD is rare, it is a common disease group that causes permanent sequelae when undiagnosed. Early detection with some skin findings is possible. In this regard, an exceptionally detailed physical examination of newborn babies becomes essential. Ultrasonography is an inexpensive and noninvasive imaging method. Evaluation of patients having suspicious skin lesions with ultrasonography will guide clinicians. The risk of major spinal dysraphism is exceedingly minimal in healthy infants with sacral dimples.

Routine physical examination of the lumbar region for cutaneous manifestations of OSD is imperative to confirm prompt treatment and avoid potentially devastating repercussions.

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

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Conflict of interest

None of the authors received any type of financial support that could be considered potential conflict of interest regarding the manuscript or its submission.

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